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Life Stories with APDS

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Learn about PI3Kδ (APDS) Activation syndrome

Sara's story

Hello everyone, my name is Sara and I'd like to tell you my story.
It's about bravery, love and resilience which shaped our family during the last years. I was born after a full-term pregnancy, but my entrance into the world was marked by complications that would have triggered concerns, aggravated by the possibility of a partial heart attack in the placenta.

In the first months of my life, after all I was fine. But, starting from the age of two, I began suffering from mucopurulent otitis, a very trublesome disorder that involved ears and eyes, that often made me ill. In spite of the taking of antibiotics prescribed by my pediatrician, my problem was still present, so I went to the hospital for further follow ups. Later I got a very aggressive stomatitis and I needed to be hospitalized.

In the meantime the follow ups and the several examinations highlighted a lymphocytes' deficiency, that explained the frequent otitis and the eye issues. I began to take immunoglobulins intravenous every 15 days to provide for this lack and, in that period, I also was subjected to adenoids' removal. **In spite of challenges and obstacles about my health, I was able to lead a relatively normal life, attending school and often travelling. T**he only waiver was swimming.

At the age of 18, I found out to have my first Hodgkin's lymphoma, followed by other two during the next years. At the beginning this disease had been identified as an unknown lymphomatous form, but later, thanks to the cooperation of Centres with branches in other european Countries, **the tests led to the suspicion of the APDS syndrome, caused by a genetic mutation de novo. Although chemiotherapy and issues tied to the lymphoma, I never lost resolution.**

After a decade of a relative stability and a switch to a subcutaneous therapy, I had to face a second lymphoma. Now, after other 5 years, I'm facing a third one, but this time the treatment has been tailored to my conditions. The medical team assessed different options, stem cells transplant included, but the final choice was a monoclonal antibodies therapy every 3 weeks.

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Although, unlike chemotherapy, the response is not immediate, **I'm facing this situation with optimism.** The total prevention of lymphomas is not possible but, thanks to recurring follow-ups, I try to manage my condition at its best. I I often have to go to the hospital for routine or extra controls, like PET, TAC or coloscopies.

Even later my path has never been without obstacles. Today, at 36, I think about my life and the faced challenges. The early menopause, due to chemiotherapies, is an issue often overlooked by doctors. My mental health has been put to a hard test and, although a private psychological support, I'm going on fighting with permanent anxiety relevant to the fact that my body could let me down at any moment.

My hometown hospital and, in particular, my immunologist, played an important role in understanding and managing my disease. I was able to live like my peers, to get a driving license, to attend university, to be indipendent. When I was a child I remember my parents very concerned, going from one doctor to another without having replies. **We fought together: this has been our strenght against a serious chronic disease. I never gave up** and certainly this influenced very much the person I'm today.

During the days in the hospital I met many other patients with Primary Immunodeficiencies; however I 've never recognized myself in their stories. Then, one day, by chance, I found one of the leaflets made by AIP: it talked about the APDS syndrome and finally I recognized myself in the words I was reading. The information collection, the involvement in their initiatives, the meetings with doctors and patients have been source of support and understanding in a path which normally was isolating.

The strenght I found in myself and my mother's resolution are a proof that it's possible to overcome the most difficult trials. I hope that our story could inspire those who face similar challenges and could show that, even in the darkest moments, the light of love can shine intensely.

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Leo's story

My name is Leo and I suffer from a very rare disease called PI3Kδ Activation syndrome, called more simply APDS. I was diagnosed when I was 3 years old; now I'm 20 and I've got a part time job.

When I was a child, I had a stunted growth, for that reason our pediatrician suggested my partents to get me tested for celiac disease. The outcomes showed an extremely low number of immunoglobulines and I was diagnosed a Primary Immunodeficiency.

Further tests showed I had very low levels of antibodies called IgG, IgA and IgE and too high levels of M (IgM) immunoglobuline. **The outcomes of my tests have been sent to labs and hospitals all over the world with the hope to get a definitive diagnoses.**

My main symptoms tend to affect intestine and lungs: these are frequent and disabling. **The treatment I'm taking is called immunoglobuline, that means that I get antibodies every three weeks.** During the years I underwent many tests and treatments: too many to be counted or mentioned.

When I was a child my mother bribed me with little prizes, candies or new toys, every time I was able to face with no tantrum a new follow-up or an umpteenth treatment. This allowed me to be distracted and braver. It sounds stupid but it worked.

I tried in many ways to lead a normal life, managing school, job, friends and medical follow-ups. I was often forced to live in an hospital for months. I'd have liked to go on studying, attending university, but my health got the better of me and I preferred to look for a job to be indipendent from my parents. Now I'm a waiter in a pizza restaurant. My health condition can leave me exhausted, drained, both mentally and physically. **However, I do my best to perform my role and to comply with the shifts they give me.**

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Leo's story

I'm the only person in my family to have APDS, that means that it wasn't transmitted genetically. Growing up with this disease made me feel alone, I felt like I couldn't talk to anyone about it. Even my sentimental life suffered about it: I felt it was wrong to give the burden of my suffering to another person.

Not many years ago, thanks to a Facebook group, **I found that there are other people affected and this makes me feel less isolated.** Since then I make an effort to tell my condition to friends and relatives, to express what I feel and to share the distresses. I explain my disease to the others in the simplest way possible: I tell them I have no anticorps so I'm inclined to contract many infections.

I'd like to meet in person other people with my same illness, to build a sort of community where we can talk and compare ourselves with each other.

I wish myself not to be stopped by the barriers tied to my illness and to be able to live my future to the fullest.

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Lucia and her father Carlo's story

My name is Carlo and I'm Lucia's dad.
My daugther suffers from a rare illness involving her immune system, the APDS syndrome. **It took 19 years to receive a definite diagnosis.** Now she is 24 and works as a secretary.

For many years she has been dealing with endless coughings and colds and she often had to be absent from school due to her infections. Every two weeks we had to go to the doctor to get a prescription for antibiotics. She was afraid to be with her peers and to play with them as she feared to contract flu, with severe consequences on breathing and hearing. She gave up with birthday parties and with the afternoons in the parish, in order to defend her health, always very weak. As you can imagine, **this pathology had a strong impact on her social and relational life.**

One day my doctor prescribed me some blood tests to verify possible reasons of a persistent cough. The outcomes revealed I got a pulmonary disease and white blood cell deficiency. The doctor informed me that this deficiency could be inherited and, following his suggestion, I had my three little girls take the tests: two of them were found to have a defective gene.

Unfortunately the final APDS diagnoses still required time and patience, also due to an unsuitable communication between doctors and hospitals. For many years Lucia has been suffering from a general illness, temperature and various infections; on some days she was so sick not to be able to get out from home.

Since when the illness has been identified and finally the suitable treatment has been adopted, her condition improved and increasingly under control. Although some infections that cannot be prevented by drugs still happen, today my daugther often feels tired, perhaps due to the many infections contracted during the years.

The life style of all my family had to be adjusted: **we started to pay attention in following a balanced diet and to adopt behaviours that support Lucia and her sister's health.**

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Since the time of diagnoses, however, there was no shortage of difficult days. Our family repeatedly deals with many obstacles and one of these is surely the lack of training of the medical staff and healthcare workers involved in my daughter's illness. It's very frustrating having to explain symtoms and conditions to those who should know about them most and should be able to address us to a right path, above all as we fear the impact that a wrong prescription could have on the treatment and the therapy.

I wish that there will be more and more training and divulgation about rare diseases such as APDS, not only for immunologists and other medical specialists, but also for paediatricians, general practitioners and all health professionals.

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